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Pilomatricoma in Children : Common but often Misdiagnosed

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Abstract. A pilomatricoma, also known as pilomatrixoma or calcifying epithelioma of Malherbe, is a benign skin tumor arising from the hair follicle matrix. This tumor is common in children and young adults, especially in the head and neck region. However, pilomatricomas are frequently misdiagnosed or not recognized. The history is typical of a slowly enlarging mass, irregularly contoured, it is fixed to the skin but slides freely over the underlying tissues, often with a discolouration which varies from red to purple-bluish. Ultrasound examination, MRI-scan and fine-needle aspiration can be helpful if the diagnosis is uncertain. Spontaneous regression has never been observed and malignant degeneration is very rare. Surgical excision with clear margins is the treatment of choice, otherwise recurrence may occur due to incomplete resection.

Introduction

Pilomatricoma, also known as calcifying epithelioma of Malherbe, is a benign skin tumor arising from the outer root sheath cell of the hair follicle. In the pediatric population, this skin neoplasm is relatively common, especially in the head and neck region (1). Despite the fact that pilomatricomas are frequent occurring skin tumors in children, that the history is typical of a slowly growing tumor and that the characteristic clinical appearance is of a superficial hard mass with discoloration, this lesion is often misdiagnosed or not recognized and confused with other skin conditions (2). The treatment however is straightforward : after complete surgical excision, recurrence is unlikely and malignant degeneration is very rare (3).

In this article, we present several cases of pilomatricomas with a classical presentation together with a brief review of the literature. We also describe an atypical case : a 1.5-year-old girl with a rapidly growing mass in the face, suggestive for a malignant tumor, in whom the final diagnosis of pilomatricoma was only made after incisional biopsy. Shortly after this surgery she developed a recurrence and complete surgical excision was performed with an uneventful postoperative follow-up.

Pilomatricomas with Typical Presentation

A 6-year old girl was referred with a slowly growing mass on the left elbow. The tumor had been present for 2 months, was adherent to the overlying skin, but mobile from underlying tissues, with a purple discoloration (Fig. 1). A pilomatricoma was suspected because of this typical appearance and complete excision was performed. Pathological examination confirmed the clinical diagnosis and the postoperative follow-up during 1 year was uneventful.

A 7-year old girl presented with a slowly growing, painless tumor on the right cheek (Fig. 2). The mass had been present for several months. Clinical examination revealed a mobile, red-bluish irregular mass with calcifications in the right preauricular region, fixed to the ulcerated overlying skin. A pilomatricoma was suspected based on the clinical signs and the lesion was excised. The pathology report confirmed the diagnosis and the postoperative follow-up during 18 months was uneventful.

A 20-year old girl was seen with a pendulous tumor at the back of the left thigh (Fig. 3). The mass had been present for 1 year, growing slowly. At clinical examination, a purple tumor was seen with irregular nodes, hard at palpation but mobile from the underlying tissues. A pilomatricoma was diagnosed because of the typical signs and excision with primary closure was performed. The clinical diagnosis was confirmed by histological examination and follow-up during 1 year was uneventful.

A 5-year old girl consulted for a mass in the right preauricular region. This had been present for 7 months and was slowly growing. A painless, hard 1.5 cm lesion was palpated, mobile from the underlying tissues with irregular calcifications (Fig. 4). Pilomatricoma was suspected and the lesion was excised ; the pathology report confirmed the clinical diagnosis. Follow-up during 15 months was uneventful.

Fig. 1
Pilomatricoma in the region of the left elbow, note the irregular nodes and purple discoloration.

Fig. 2
Pilomatricoma on the right cheek, note the ulcerated overlying skin and irregular nodules with calcifications.

Pilomatricoma with Unusual Presentation

A 1.5-year-old girl was referred to our department with a rapidly growing, painless tumor on the right cheek. This lesion was present for 7 months, starting as a small mole. No lesion was noticed at birth. Apart form this lesion the child was perfectly healthy. The general practitioner suspected a hematoma and performed a puncture, after which a rapid growth was observed with purple-red dis-

Fig. 3
Pilomatricoma on the back of the left thigh, note the thin overlying skin, purple discoloration and irregular nodes.

Fig. 4
Pilomatricoma in the right preauricular region, note irregular nodules and thin skin.

Fig. 5
Pilomatricoma on the right cheek, note the purple red color. No calcifications or nodules can be seen, as opposed to the other pilomatricomas shown previously.

Fig. 6
Six weeks after incisional biopsy, recurrence and rapid growth was observed. Calcifications can be seen at this stage and the lesion has more typical characteristics of a pilomatricoma.

coloration. At clinical examination, a purple-red colored, firm mass was noted on the right cheek, measuring 2,5 cm. The tumor was mobile but the borders were not well defined. The overlying skin was very thin and in danger of being perforated (Fig. 5). Ultrasound examination showed a well-demarcated, hypo-echogenic nodule. Due to the rapid growth and the possibility of a malignancy, a MRI-scan was performed, which showed a sharply demarcated, contrast fixing cystic lesion in the subcutis of the cheek. No aspiration was performed. To obtain a diagnosis and to decrease the contents of the mass, an incisional biopsy was performed with frozen section, which showed a ruptured cyst and pilomatricoma on final analysis. A few weeks later, the parents returned because of tumor recurrence with rapid growth (Fig. 6). A complete surgical resection with primary closure was performed. Pathological examination showed a pilomatricoma, completely resected with free margins. Follow-up during 10 months was uneventful with no recurrence (Fig. 7).

Fig. 7
Six months postoperative view after complete surgical excision, no signs of recurrence.

Discussion

Biological description of the lesion

Pilomatricoma, also known as pilomatrixoma or calcifying epithelioma, was first described by Malherbe and

Chenantais in 1880 (4). They thought the lesion originated from the sebaceous glands and named it calcifying epithelioma, due to the calcifications often found in these lesions. Dubreuilh and Cazenave described the characteristic histology of this lesion, including islands of epithelial cells and shadow cells (5). The tumor represents a disturbance of the hair follicle cycle, where final development toward mature hair with matrix, cortex and inner hair sheath fails to take place. The terms pilomatrixoma and pilomatricoma were proposed later due to their origin from the hair matrix cells and to avoid confusion with malignancy, as this is a benign lesion (6).

The tumor accounts for less than 2% of all primary cutaneous neoplasms and less than 0.5% of cases in an active dermatopathologic practice. More than half of the tumors are diagnosed in the first two decades of life and most of these occur in children younger than 10 years of age with a female preponderance. In fact, pilomatricomas are one of the most common solid cutaneous tumors in patients of 20 years or younger (1, 7, 8). The most frequent location is the head and neck region, especially the peri-orbital, lateral cheek and pre-auricular areas (8-13). Most pilomatricomas are solitary lesions. Multiple or recurring lesions are rare and have been associated with certain syndromes such as myotonic dystrophy or Gardner's syndrome, which should be screened by the physician in these cases (2).

Spontaneous regression is never observed and malignant degeneration is extremely rare (3, 8, 12). Complete surgical excision including the overlying ulcerated skin is the treatment of choice. After complete resection, recurrence rate is zero ; if there is a recurrence, it proves that the excision was not complete. Histological examination reveals well-circumscribed nodules in the subcutaneous tissues, surrounded by a connective tissue capsule. Two distinct cell populations are visible : shadow or ghost cells, which have a central unstained area of lost nuclei, surrounded by basaloid cells. Calcium deposits are frequently found, giving the lesion the rock-like feeling on palpation (9).

Differential diagnosis

The differential diagnosis for pilomatricomas is heterogenous. They may be mistaken for epidermoid or dermoid cysts, calcified lymph nodes, calcified hematomas, hemangiomas or parotid gland tumors. Pilomatricomas are slowly growing tumors and calcifications can often be palpated. Epidermoid and dermoid cysts on the contrary, are characterized by normal overlying skin without discoloration, which can be moved freely, they are soft masses and do not present with calcifications or irregular nodes on the skin. Therefore these conditions are easily differentiated from pilomatricomas.

Pain or tenderness is uncommon in pilomatricomas and although previous trauma is sometimes described, causality is often not clear. A red to purple-bluish discoloration is often seen and this is very characteristic. As mentioned, these tumors can occur at any age, but children and young adolescents are most frequently affected with a female preponderance as seen in our five patients.

Although the characteristics of pilomatricoma are very typical and pilomatricomas are common in children, the preoperative diagnosis is often missed or confused with other skin conditions even by experienced physicians or surgeons. In various studies, a correct preoperative diagnosis was only made in 30 to 50% of the cases. The most common misdiagnoses were dermoid cyst, sebaceous cyst and unidentified masses (3, 9, 12).

In patients with uncommon clinical features, such as multicentricity, large size or deep location or abnormal colour, confusion may arise concerning diagnosis. In these cases or when a malignancy is suspected, ultrasound examination, computed tomography or MRI-scan can be performed, combined with fine-needle aspiration (12, 14). Ultrasound examination can demonstrate the superficial position, the continuity of the lesion with deeper structures and the degree of calcification. It is a low-cost, non-invasive tool and more appropriate for younger children, because there is no need for sedation or anesthesia. CT or MRI-scans can reveal a well-described, soft tissue mass, with or without visible calcifications and can be valuable for lesions located in the parotid region as these can be mistaken for parotid neoplasms and to differentiate from lymph nodes (12, 13). Fine-needle aspiration may reveal the presence of characteristic ghost cells and basaloid cells. If these cells are not present, the cytological diagnosis may be misleading and be confused with malignancy (14).

The first four cases presented with a lesion, which had the typical clinical features and history of a pilomatricoma : a slowly growing, painless, irregularly contoured hard mass, fixed to the overlying skin, but mobile from the subcutaneous tissues with discoloration and palpable calcifications. The lesions were easily and correctly diagnosed, without additional investigations.

In our last described case, neither the history nor the clinical features were typical. As mentioned in the literature, these aspects were the reasons the tumor was misdiagnosed and not recognized. The lesion had grown very fast in a few months, so a malignancy or a vascular tumor were suspected. A trauma was present (puncture by the general practitioner), after which rapid growth of the tumor was observed, thus masking the correct diagnosis. The clinical presentation was very uncommon, as calcifications were not present and the lesion was very red and firm (not hard) in palpation, so none of the three examining experienced surgeons had even considered the tumor as being a pilomatricoma. Imaging provided no extra information and in fact was misleading. On ultrasonography the lesion was diagnosed as a heman-

gioma and on MRI-scan as a lymphatic malformation by the radiologist. For practical reasons, fine needle aspiration was not possible. To obtain a diagnosis, an incisional biopsy was done, which showed pilomatricoma in the final histology report. As mentioned, recurrence is very likely after incomplete resection and a rapid recurrence of the tumor after incisional biopsy was indeed observed. Complete surgical resection provided definite treatment with uneventful follow-up.

The photographs presented in this article also show that the clinical aspects of a pilomatricoma can be very variable and in our opinion, this has not been so clearly demonstrated in the literature concerning pilomatricomas.

In conclusion, pilomatricoma is a frequent occurring benign tumor in the pediatric population, especially in the head and neck region. In most cases, the clinical presentation is quite typical and the diagnosis is obvious. Despite these facts, pilomatricomas are often misdiagnosed even by experienced physicians, especially in cases where the clinical presentation is unusual. Surgery is the preferred treatment**.**

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